

38 Ashkenazi genetic diseases
1. Bloom Syndrome ( 1/134 )
2. Canavan Disease ( 1/55 )
3. Cystic Fibrosis ( 1/24 )
4. Familial Dysautonomia ( 1/31 )
5. Familial Hyperinsulinism ( 1/68 )
6. Fanconi Anemia C ( 1/100 )
7. Gaucher Disease ( 1/15 )
8. Glycogen Storage Disease 1A ( 1/64 )
9. Joubert Syndrome 2 ( 1/110 )
10. Lipoamide Dehydrogenase Deficiency (E3) ( 1/107 )
11. Maple Syrup Urine Disease 1B ( 1/97 )
12. Mucopolidosis IV ( 1/89 )
13. Nemaline Myopathy ( 1/168 )
14. Niemann-Pick Disease ( 1/115 )
15. Spinal Muscular Atrophy ( 1/41 )
16. Tay-Sachs Disease ( 1/27 )
17. Usher IF ( 1/147 )
18. Usher III ( 1/120 )
19. Walker Warburg ( 1/120 )
20. 3-Phosphoglycerate Dehydrogenase Deficiency ( 1/280 )
21. Abetalipoproteinemia ( 1/180 )
22. Alport Syndrome, Autosomal Recessive ( 1/188 )
23. Arthrogryposis, Mental Retardation and Seizures ( 1/373 )
24. Bardet-Biedl Syndrome ( 1/107 )
25. Carnitine Palmitoyltransferase II Deficiency ( 1/51 )
26. Congenital Amegakaryocytic Thrombocytopenia ( 1/55 )
27. Congenital Disorder of Glycosylation Ia ( 1/57 )
28. Dyskeratosis Congenita, Autosomal Recessive ( 1/203 )
29. Ehlers-Danlos VIIIC ( 1/248 )
30. Fragile X Syndrome ( 1/115 )
31. Galactosemia ( 1/172 )
32. Multiple Sulphatase Deficiency ( 1/320 )
33. Polycystic Kidney Disease, Autosomal Recessive ( 1/107 )
34. Retinitis Pigmentosa 59 ( 1/118 )
35. Smith-Lemli-Opitz Syndrome ( 1/36 )
36. Tyrosinemia I ( 1/150 )
37. Wilson Disease ( 1/70 )
38. Zellweger Syndrome ( 1/172 )